# Tyrosinemias (TYR-I, TYR-II)*

An amino acid disorder

***What is it?***

Tyrosinemias, (also known as TYR-I and TYR-II) are an inherited amino acid disorder. People with amino acid disorders, like TYR-I and TYR-II, cannot properly break down certain components of protein. This is because the body is lacking a specific chemical called an enzyme. Since the body cannot properly break down the protein, certain amino acids build up in the blood and urine and cause problems when a person eats normal amounts of protein.

***What are the symptoms?***

People with TYR-I and TYR-II appear normal at birth. If left untreated, people with TYR-I and TYR-II will develop eye problems, blisters on the hands and feet, developmental delays, and behavioral problems. Many symptoms of TYR-I and TYR-II can be prevented by immediate treatment and lifelong management. People with TYR-I and TYR-II typically receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

***What is the treatment?***

TYR-I and TYR-II are treated by eating a diet low in protein. People with TYR-I and TYR-II are given a special formula that is low in certain components of protein. People with TYR-I and TYR-II typically receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

***Inheritance and frequency***

TYR-I and TYR-II are inherited in an autosomal recessive manner. This means that for a person to be affected with TYR-I and TYR-II, he or she must have inherited two non-working copies of the gene responsible for causing TYR-I and TYR-II. Usually, both parents of a person affected with an autosomal recessive disorder are unaffected because they are carriers. This means that they have one working copy of the gene, and one non-working copy of the gene. When both parents are carriers, there is a 1 in 4 (or 25%) chance that both parents will pass on the non working copies of their gene, causing the baby to have TYR-II or TYR-II. Typically, there is no family history of TYR-I or TYR-II in an affected person. TYR-I and TYR-II are rare amino acid disorder; the number of people with TYR-I or TYR-II is not known.

***How is it detected?***

TYR-I and TYR-II may be detected through newborn screening. A recognizable pattern of elevated chemicals alerts the laboratory that a baby may be affected. Confirmation of newborn screening results is required to make a firm diagnosis. This is usually done by a physician that specializes in metabolic conditions, or a primary care physician.

***How is it treated?***

TYR-I and TYR-II are treated by eating a diet low in protein and drinking a special formula as recommended by a genetic metabolic medical professional.

***There is a lower probability of detection of this disorder during the immediate newborn period**.

**DISCLAIMER: This information is not intended to replace the advice of a genetic metabolic medical professional.**

## For more information:

**Genetics Home Reference**
Website: http://www.ghr.nlm.nih.gov

**National Coalition of PKU and other allied disorders**
PO Box 1244
Mansfield, MA 02048
Phone: 877-996-2723
Website: http://www.PKU-allieddisorders.org/

**Save Babies Through Screening Foundation**
4 Manor View Circle
Malvern, PA 19355-1622
Toll Free Phone: 1-888-454-3383
Fax: (610) 993-0545
Email: email@savebabies.org
Website: http://www.savebabies.org

**STAR-G Hawaii Department of Health**
http://www.newbornscreening.info/parents/aminoaciddisorders/ASAL.html